



critical congenital heart disease

Critical congenital heart disease (CCHD) is a term that refers to a group of serious heart defects that are present from birth. These abnormalities result from problems with the formation of one or more parts of the heart during the early stages of embryonic development. CCHD prevents the heart from pumping blood effectively or reduces the amount of oxygen in the blood. As a result, organs and tissues throughout the body do not receive enough oxygen, which can lead to organ damage and life-threatening complications. Individuals with CCHD usually require surgery soon after birth.

Although babies with CCHD may appear healthy for the first few hours or days of life, signs and symptoms soon become apparent. These can include an abnormal heart sound during a heartbeat (heart murmur), rapid breathing (tachypnea), low blood pressure (hypotension), low levels of oxygen in the blood (hypoxemia), and a blue or purple tint to the skin caused by a shortage of oxygen (cyanosis). If untreated, CCHD can lead to shock, coma, and death. However, most people with CCHD now survive past infancy due to improvements in early detection, diagnosis, and treatment.

Some people with treated CCHD have few related health problems later in life. However, long-term effects of CCHD can include delayed development and reduced stamina during exercise. Adults with these heart defects have an increased risk of abnormal heart rhythms, heart failure, sudden cardiac arrest, stroke, and premature death.

Each of the heart defects associated with CCHD affects the flow of blood into, out of, or through the heart. Some of the heart defects involve structures within the heart itself, such as the two lower chambers of the heart (the ventricles) or the valves that control blood flow through the heart. Others affect the structure of the large blood vessels leading into and out of the heart (including the aorta and pulmonary artery). Still others involve a combination of these structural abnormalities.

People with CCHD have one or more specific heart defects. The heart defects classified as CCHD include coarctation of the aorta, double-outlet right ventricle, D-transposition of the great arteries, Ebstein anomaly, hypoplastic left heart syndrome, interrupted aortic arch, pulmonary atresia with intact septum, single ventricle, total anomalous pulmonary venous connection, tetralogy of Fallot, tricuspid atresia, and truncus arteriosus.

Frequency

Heart defects are the most common type of birth defect, accounting for more than 30 percent of all infant deaths due to birth defects. CCHD represents some of the most

serious types of heart defects. About 7,200 newborns, or 18 per 10,000, in the United States are diagnosed with CCHD each year.

Genetic Changes

In most cases, the cause of CCHD is unknown. A variety of genetic and environmental factors likely contribute to this complex condition.

Changes in single genes have been associated with CCHD. Studies suggest that these genes are involved in normal heart development before birth. Most of the identified mutations reduce the amount or function of the protein that is produced from a specific gene, which likely impairs the normal formation of structures in the heart. Studies have also suggested that having more or fewer copies of particular genes compared with other people, a phenomenon known as copy number variation, may play a role in CCHD. However, it is unclear whether genes affected by copy number variation are involved in heart development and how having missing or extra copies of those genes could lead to heart defects. Researchers believe that single-gene mutations and copy number variation account for a relatively small percentage of all CCHD.

CCHD is usually isolated, which means it occurs alone (without signs and symptoms affecting other parts of the body). However, the heart defects associated with CCHD can also occur as part of genetic syndromes that have additional features. Some of these genetic conditions, such as Down syndrome, Turner syndrome, and 22q11.2 deletion syndrome, result from changes in the number or structure of particular chromosomes. Other conditions, including Noonan syndrome and Alagille syndrome, result from mutations in single genes.

Environmental factors may also contribute to the development of CCHD. Potential risk factors that have been studied include exposure to certain chemicals or drugs before birth, viral infections (such as rubella and influenza) that occur during pregnancy, and other maternal illnesses including diabetes and phenylketonuria. Although researchers are examining risk factors that may be associated with this complex condition, many of these factors remain unknown.

Inheritance Pattern

Most cases of CCHD are sporadic, which means they occur in people with no history of the disorder in their family. However, close relatives (such as siblings) of people with CCHD may have an increased risk of being born with a heart defect compared with people in the general population.

Other Names for This Condition

- CCHD
- critical congenital heart defects

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Congenital heart disease
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0152021/>
- Genetic Testing Registry: Ebstein's anomaly
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0013481/>
- Genetic Testing Registry: Hypoplastic left heart syndrome
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0152101/>
- Genetic Testing Registry: Hypoplastic left heart syndrome 2
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C3280795/>
- Genetic Testing Registry: Persistent truncus arteriosus
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0041207/>
- Genetic Testing Registry: Pulmonary atresia with intact ventricular septum
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0344975/>
- Genetic Testing Registry: Pulmonary atresia with ventricular septal defect
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0344976/>
- Genetic Testing Registry: Tetralogy of Fallot
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0039685/>
- Genetic Testing Registry: Transposition of the great arteries
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0040761/>
- Genetic Testing Registry: Transposition of the great arteries, dextro-looped 2
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C3151220/>
- Genetic Testing Registry: Transposition of the great arteries, dextro-looped 3
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C3151221/>
- Genetic Testing Registry: Tricuspid atresia
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0243002/>

Other Diagnosis and Management Resources

- Baby's First Test: Critical Congenital Heart Disease
<http://www.babysfirsttest.org/newborn-screening/conditions/critical-congenital-heart-disease-cchd>
- Boston Children's Hospital
<http://www.childrenshospital.org/conditions-and-treatments/conditions/congenital-heart-defects>

- Centers for Disease Control and Prevention: Screening for Critical Congenital Heart Defects
<https://www.cdc.gov/ncbddd/heartdefects/cchd-facts.html>
- Children's Hospital of Philadelphia
<http://www.chop.edu/conditions-diseases/congenital-heart-disease#.VE49mcIn19Y>
- Cincinnati Children's Hospital Medical Center
<https://www.cincinnatichildrens.org/patients/child/encyclopedia/defects>
- Cleveland Clinic
<http://my.clevelandclinic.org/health/articles/congenital-heart-disease>
- Screening, Technology, and Research in Genetics (STAR-G)
<http://www.newbornscreening.info/Parents/otherdisorders/CCHD.html>
- University of California, San Francisco Fetal Treatment Center: Congenital Heart Disease
<http://fetus.ucsf.edu/congenital-heart-disease>

General Information from MedlinePlus

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>

Additional Information & Resources

MedlinePlus

- Encyclopedia: Coarctation of the Aorta
<https://medlineplus.gov/ency/article/000191.htm>
- Encyclopedia: Double Outlet Right Ventricle
<https://medlineplus.gov/ency/article/007328.htm>
- Encyclopedia: Ebstein's Anomaly
<https://medlineplus.gov/ency/article/007321.htm>
- Encyclopedia: Hypoplastic Left Heart Syndrome
<https://medlineplus.gov/ency/article/001106.htm>

- Encyclopedia: Pulmonary Atresia
<https://medlineplus.gov/ency/article/001091.htm>
- Encyclopedia: Tetralogy of Fallot
<https://medlineplus.gov/ency/article/001567.htm>
- Encyclopedia: Total Anomalous Pulmonary Venous Return
<https://medlineplus.gov/ency/article/001115.htm>
- Encyclopedia: Transposition of the Great Vessels
<https://medlineplus.gov/ency/article/001568.htm>
- Encyclopedia: Tricuspid Atresia
<https://medlineplus.gov/ency/article/001110.htm>
- Encyclopedia: Truncus Arteriosus
<https://medlineplus.gov/ency/article/001111.htm>
- Health Topic: Congenital Heart Defects
<https://medlineplus.gov/congenitalheartdefects.html>
- Health Topic: Newborn Screening
<https://medlineplus.gov/newbornscreening.html>

Genetic and Rare Diseases Information Center

- Hypoplastic left heart syndrome
<https://rarediseases.info.nih.gov/diseases/6739/hypoplastic-left-heart-syndrome>
- Transposition of the great arteries
<https://rarediseases.info.nih.gov/diseases/7795/transposition-of-the-great-arteries>

Additional NIH Resources

- National Heart Lung and Blood Institute: What Is Tetralogy of Fallot?
<https://www.nhlbi.nih.gov/health/health-topics/topics/tof/>

Educational Resources

- Centers for Disease Control and Prevention: Congenital Heart Defects
<https://www.cdc.gov/ncbddd/heartdefects/>
- Disease InfoSearch: Congenital Heart Defects
<http://www.diseaseinfosearch.org/Congenital+Heart+Defects/1833>
- Health Resources and Services Administration: Critical Congenital Heart Disease Added to Recommended Uniform Screening Panel
<https://www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders/recommendations/correspondence/cyanoticheartsecre09212011.pdf>
- KidsHealth from Nemours
<http://kidshealth.org/en/parents/congenital-heart-defects.html>

- Lucile Packard Children's Hospital at Stanford
<http://www.stanfordchildrens.org/en/topic/default?id=factors-contributing-to-congenital-heart-disease-90-P01788>
- MalaCards: critical congenital heart disease
http://www.malacards.org/card/critical_congenital_heart_disease
- March of Dimes
<http://www.marchofdimes.org/baby/congenital-heart-defects.aspx>
- Merck Manual Consumer Version
<http://www.merckmanuals.com/home/children-s-health-issues/birth-defects/heart-defects>
- Orphanet: Hypoplastic left heart syndrome
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=2248
- Orphanet: Pulmonary atresia-intact ventricular septum syndrome
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=1208
- Orphanet: Pulmonary atresia with ventricular septal defect
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=1207
- Orphanet: Tetralogy of Fallot
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=3303
- Orphanet: Tricuspid atresia
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=1209
- Orphanet: Truncus arteriosus
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=3384

Patient Support and Advocacy Resources

- Adult Congenital Heart Association
<https://www.achaheart.org/>
- American Heart Association
http://www.heart.org/HEARTORG/Conditions/CongenitalHeartDefects/Congenital-Heart-Defects_UCM_001090_SubHomePage.jsp
- Contact a Family (UK)
<http://www.cafamily.org.uk/medical-information/conditions/h/heart-defects/>
- Resource List from the University of Kansas Medical Center
<http://www.kumc.edu/gec/support/conghart.html>

ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%22critical+congenital+heart+disease%22+OR+%22critical+congenital+heart+defects%22>

Scientific Articles on PubMed

- PubMed
https://www.ncbi.nlm.nih.gov/pubmed?term=%28Heart+Defects,+Congenital%5BMAJR%5D%29+AND+%28%28critical+congenital+heart+disease%5BTIAB%5D%29+OR+%28critical+congenital+heart+defect*%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- COARCTATION OF AORTA
<http://omim.org/entry/120000>
- CONOTRUNCAL HEART MALFORMATIONS
<http://omim.org/entry/217095>
- EBSTEIN ANOMALY
<http://omim.org/entry/224700>
- HETEROTAXY, VISCERAL, 2, AUTOSOMAL
<http://omim.org/entry/605376>
- HYPOPLASTIC LEFT HEART SYNDROME 1
<http://omim.org/entry/241550>
- HYPOPLASTIC LEFT HEART SYNDROME 2
<http://omim.org/entry/614435>
- PULMONARY ATRESIA WITH INTACT VENTRICULAR SEPTUM
<http://omim.org/entry/265150>
- PULMONARY ATRESIA WITH VENTRICULAR SEPTAL DEFECT
<http://omim.org/entry/178370>
- TETRALOGY OF FALLOT
<http://omim.org/entry/187500>
- TOTAL ANOMALOUS PULMONARY VENOUS RETURN 1
<http://omim.org/entry/106700>
- TRANSPOSITION OF THE GREAT ARTERIES, DEXTRO-LOOPED 1
<http://omim.org/entry/608808>
- TRANSPOSITION OF THE GREAT ARTERIES, DEXTRO-LOOPED 3
<http://omim.org/entry/613854>
- TRICUSPID ATRESIA
<http://omim.org/entry/605067>

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